2018 KABUKI SYNDROME RESEARCH CONFERENCE SPEAKER BIOS

DR. SIDDHARTH BANKA – UNIVERSITY OF MANCHESTER, U.K.

Dr. Banka is a Clinician Scientist working at the Division of Evolution and Genomic Sciences (University of Manchester). He holds a joint position at the Manchester Centre for Genomic Medicine (St Mary's Hospital, Manchester University NHS Trust) as a Consultant Clinical Geneticist.

Dr. Banka's research group uses a combination of genomics, clinical and functional studies to (1) identify novel disease-genes and mechanisms; (2) improve interpretation of genomic data; and (3) delineate the phenotypic spectrum of rare gene diseases.

His current research programs are primarily around the pediatric disorders of chromatin remodeling, conditions caused by copy number variations and inborn errors of metabolism. He is the Research coordinator for the European Reference Network for Intellectual Disabilities and Congenital Malformations (ERN-ITHACA). He established and runs the Chromatin Disorders Clinic at the Manchester Centre for Genomic Medicine. He is also the Pathway lead for the MSc for the NHS National Genomics Scientist Training Program (STP).

Dr. Banka graduated in Medicine (MBBS) from Lokmanya Tilak Medical College (University of Mumbai) in 2002, obtained Membership of Royal College of Pediatrics and Child Health (MRCPCH) in 2006 and joined the Clinical Genetics training program in Manchester in 2007. He was awarded a Research Training Fellowship by the NIHR Manchester Biomedical Research Centre from 2008 to 2011 and a PhD by the University of Manchester in 2012. He obtained his Certificate of Completion of Specialist Training (CCST) in Clinical Genetics training in 2013. Dr. Banka has been in his current position since July 2013.

DR. JACQUELINE HARRIS – KENNEDY KRIEGER INSTITUTE

Dr. Jacqueline Harris (nee Weissman) is the director of the Tuberous Sclerosis Clinic at Kennedy Krieger Institute and an assistant professor in neurology at Johns Hopkins Hospital.

Dr. Harris received her BA with Honors in English Language and Literature and Neurobiology from The University of Chicago. She earned an MD from Cleveland Clinic Lerner College of Medicine of Case Western Reserve University and an MS in Clinical Research Scholars Program from Case Western Reserve University.

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She completed a residency in pediatrics at the Children's Medical Center at UT Southwestern.

Dr. Harris was chief resident in the Neurodevelopmental Disabilities Program at Johns Hopkins Hospital through 2016. She is currently the director of the Tuberous Sclerosis and Related Disorders Clinic at Kennedy Krieger Institute, an attending in the Rehabilitation Unite and Neurology and Neurogenetics Clinic, and a Neurology/Medical consultant to the Neurobehavioral Unit at Kennedy Krieger Institute.

Dr. Harris is interested in genetic and epigenetic causes of neurdevelopmental disorders - particularly intellectual disability - and how specific genetic and epigenetic changes lead to specific neuroanatomic, neurophysiologic and cognitive phenotypes. She is also interested in developing specific cognitive profiles in genetic syndromes as potential outcome measures for trails and to help localize deficits. Most of her research is currently centered around Kabuki syndrome.

DR. HANS BJORNSSON – JOHNS HOPKINS UNIVERSITY SCHOOL OF MEDICINE

Dr. Hans Tomas Bjornsson is an assistant professor of pediatrics and genetics at the Johns Hopkins University School of Medicine. His areas of clinical expertise include clinical genetics and pediatrics.

He is a faculty member in the McKusick-Nathans Institute of Genetic Medicine. Dr. Bjornsson serves as the director of the Epigenetic and Chromatin Clinic and assistant program director of Clinical Genetics.

He earned his M.D. from the University of Iceland and his Ph.D. from Johns Hopkins. He completed a combined pediatrics and medical genetics residency program at Johns Hopkins.

Dr. Bjornsson’s research focuses on exploring the epigenomic impact of various Mendelian disorders of the epigenetic machinery. He is also interested in epigenetic-based therapeutic development with focus on developing therapies for Mendelian disorders of the histone machinery and imprinting disorders.

Dr. Bjornsson serves on the faculty of the Medical Scientist Training/MD-PhD Program. In 2013, he receive an Early Independence Award from the National Institutes of Health, a prestigious grant that gives promising junior scientists the opportunity to skip traditional post-doctoral training and move directly into independent research roles. He is a member of the American Academy of Pediatrics and the American Society of Human Genetics.

DR. JILL FAHRNER – JOHNS HOPKINS UNIVERSITY SCHOOL OF MEDICINE

Dr. Jill A. Fahrner is a physician scientist with a long-standing interest in epigenetics. She did her PhD research with Dr. Stephen B. Baylin in the Sidney Kimmel Comprehensive Cancer Center at Johns Hopkins. Her graduate work culminated in multiple publications in the field of cancer epigenetics. She completed medical school at the University of North Carolina and Pediatrics residency at Duke University Medical Center.

She joined the McKusick-Nathans Institute of Genetic Medicine as a Genetic Medicine resident in 2009 and completed her training in 2012. She stayed on as chief resident from 2012-2013 and then joined the faculty as an Assistant Professor in the Department of Pediatrics within the McKusick-Nathans Institute of Genetic Medicine in 2013. She has received a Johns Hopkins School of Medicine Clinician Scientist Award, a Johns Hopkins School of Medicine Musculoskeletal Pilot and Feasibility Award, and the Margaret Ellen Nielsen Fellowship Award while at Johns Hopkins. She is a 2016 Hartwell Foundation Individual Biomedical Research Award recipient.

Dr. Fahrner has clinical expertise in epigenetic disorders involving abnormal growth, including Kabuki syndrome and others. As a clinical geneticist, she helped build the Johns Hopkins Epigenetics and Chromatin clinic and has been caring for patients in this clinic since 2013. Her laboratory research focuses on mechanisms of abnormal growth in Mendelian disorders of the epigenetic machinery, particularly Kabuki syndrome and Weaver syndrome, as well as the development of novel epigenetic therapies to treat these disorders.

DR. ANDREW LINDSLEY, MD PhD – CINNCINNATI CHILDREN'S HOSPITAL

SUBSPECIALTY: Pediatric Allergy & Immunology

ACADEMIC APPOINTMENT: Assistant Professor of Pediatrics, University of Cincinnati School of Medicine

CLINICAL APPOINTMENT: Assistant Professor & Attending Physician, Division of Allergy and Immunology, Cincinnati Children's Hospital Medical Center

BACKGROUND:
Dr. Andrew Lindsley completed his undergraduate studies at Princeton University and then entered the medical scientist training program at Indiana University School of Medicine. He completed his early scientific training in the cardiovascular development lab of Simon Conway Ph.D. at the Wells Center for Pediatric Research, earning a Ph.D. by dissecting the molecular mechanisms driving persistent truncus arteriosis in the Splotch2H mouse model.

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After graduating from medical school, Dr. Lindsley continued his clinical training in the Categorical Pediatric Residency program at Cincinnati Children's Hospital Medical Center (CCHMC) and participated in the American Academy of Pediatrics–sponsored Integrated Research Pathway.

After completing residency, he continued his medical training as an Allergy and Immunology Clinical Fellow and provided care for many pediatric patients with rare immune deficiencies via the high-volume Primary Immune Deficiency clinic at CCHMC. During fellowship, Dr. Lindsley recruited and immune characterized a cohort of type 1 Kabuki syndrome patients (KMT2D/MLL2) and discovered that many KS patients have humoral immune deficiency (difficulty making protective antibodies) as well defects in B cell differentiation.

RESEARCH INTERESTS:
Dr Lindsley's translation medicine research laboratory investigates the mechanisms driving humoral immune deficiencies, with a primary focus on defects in terminal B cell differentiation. Specific pathways of interest include the non-canonical NF-kB pathway and the role of epigenetic signaling in B cell / plasmablast terminal differentiation. A major focus of Dr. Lindsley's lab involves characterizing the humoral defects associated with Kabuki syndrome, a rare disorder secondary to mutations in the epigenetic master-regulatory genes KMT2D/MLL2 and KDM6A/UTX.

In addition, Dr Lindsley collaborates with the CCHMC Division of Human Genetics in an on-going effort to utilize next generation exomic / genomic sequencing to help identify causal mutations in cases of idiopathic immune deficiency. This collaboration has identified novel NFKB2 mutations in patients with common variable immune deficiency (CVID).

Overall, the primary goal of Dr. Lindsley's research is to develop new diagnostic tools and treatment options for the management of humoral immune deficiency.